### (19) World Intellectual Property Organization

International Bureau



## - 1 1881 | 1 1888 | 1 1888 | 1 1888 | 1 1888 | 1 1888 | 1 1888 | 1 1888 | 1 1888 | 1 1888 | 1 1888 | 1 1888 |

# (43) International Publication Date 10 March 2005 (10.03.2005)

#### **PCT**

# (10) International Publication Number WO 2005/021787 A3

(51) International Patent Classification<sup>7</sup>: C12Q 1/48, C12N 9/12, C07K 16/00, C12N 15/00, A61K 35/00, G01N 33/00, C12N 5/00

(21) International Application Number:

PCT/IB2004/002953

(22) International Filing Date: 27 August 2004 (27.08.2004)

(25) Filing Language:

English

(26) Publication Language:

English

(30) Priority Data: 60/498,655

29 August 2003 (29.08.2003) U

(71) Applicant (for all designated States except US): INTE-GRAGEN [FR/FR]; Pépinière Génopôle Industries, CCI Essonne, 4 Rue Pierre Fontaine, F-91000 EVRY (FR).

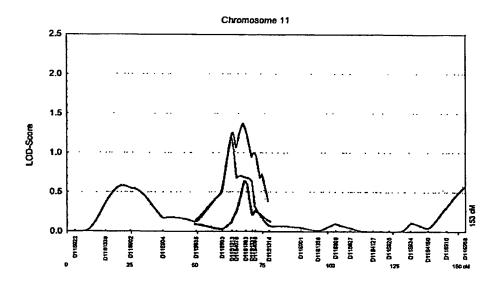
- (72) Inventors; and
- (75) Inventors/Applicants (for US only): ROSCHMANN.

Elke [DE/FR]; 32 rue Oberkampf, F-91100 Corbeil Essonne (FR). ROUSSEAU, Francis [FR/FR]; 11, rue Théophile Gautier, F-91600 Savignay sur Orge (FR).

- (74) Agent: BECKER, Philippe; Cabinet Becker et Associés, 25 Rue Louis Le Grand, F-75002 Paris (FR).
- (81) Designated States (unless otherwise indicated, for every kind of national protection available): AE, AG, AL, AM, AT, AU, AZ, BA, BB, BG, BR, BW, BY, BZ, CA, CH, CN, CO, CR, CU, CZ, DE, DK, DM, DZ, EC, EE, EG, ES, FI, GB, GD, GE, GH, GM, HR, HU, ID, IL, IN, IS, JP, KE, KG, KP, KR, KZ, LC, LK, LR, LS, LT, LU, LV, MA, MD, MG, MK, MN, MW, MX, MZ, NA, NI, NO, NZ, OM, PG, PH, PL, PT, RO, RU, SC, SD, SE, SG, SK, SL, SY, TJ, TM, TN, TR, TT, TZ, UA, UG, US, UZ, VC, VN, YU, ZA, ZM, ZW.
- (84) Designated States (unless otherwise indicated, for every kind of regional protection available): ARIPO (BW, GH, GM, KE, LS, MW, MZ, NA, SD, SL, SZ, TZ, UG, ZM, ZW), Eurasian (AM, AZ, BY, KG, KZ, MD, RU, TJ, TM), European (AT, BE, BG, CH, CY, CZ, DE, DK, EE, ES, FI, FR, GB, GR, HU, IE, IT, LU, MC, NL, PL, PT, RO, SE, SI,

[Continued on next page]

#### (54) Title: HUMAN OBESITY SUSCEPTIBILITY GENE AND USES THEREOF



(57) Abstract: The present invention discloses the identification of a human obesity susceptibility gene, which can be used for the diagnosis, prevention and treatment of obesity and related disorders, as well as for the screening of therapeutically active drugs. The invention more specifically discloses that the MAP3K11 gene on chromosome 11 and certain alleles thereof are related to susceptibility to obesity and represent novel targets for therapeutic intervention. The present invention relates to particular mutations in the MAP3K11 gene and expression products, as well as to diagnostic tools and kits based on these mutations. The invention can be used in the diagnosis of predisposition to, detection, prevention and/or treatment of coronary heart disease and metabolic disorders, including hypoalphalipoproteinemia, familial combined hyperlipidemia, insulin resistant syndrome X or multiple metabolic disorder, coronary artery disease, diabetes and dyslipidemic hypertension.



## WO 2005/021787 A3



SK, TR), OAPI (BF, BJ, CF, CG, CI, CM, GA, GN, GQ, GW, ML, MR, NE, SN, TD, TG).

Published:

- with international search report
- before the expiration of the time limit for amending the claims and to be republished in the event of receipt of amendments
- (88) Date of publication of the international search report: 14 July 2005

For two-letter codes and other abbreviations, refer to the "Guidance Notes on Codes and Abbreviations" appearing at the beginning of each regular issue of the PCT Gazette.